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Prenatal Tests

Tests that can check for certain birth defects in your unborn baby

911/303

Dear Mother-To-Be:

Congratulations! Pregnancy is usually a good experience and a time to enjoy.

Most babies are born healthy, but sometimes things can go wrong, even if you do all the right things during pregnancy. The unborn baby's major parts are formed in the first few months. If something goes wrong during this time, the baby can be born with a birth defect or other health problem.

There are tests that can check for some of these problems even before your baby is born. This booklet will tell you about three tests that can check your unborn baby's health. They can be done when you are about 16 weeks pregnant.

Of course, it's up to you to decide if you want any of these tests. Before you make up your mind, it's important that you learn as much as you can about:

- *your chances of having a baby with a birth defect or other health problem;*
- *the tests you can get before your baby is born; and*
- *what choices you have if a test finds a problem.*

This booklet can get you started. After you read it, your doctor, nurse, midwife, or a genetic counselor can tell you even more to help you make the decision that's best for you.

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Birth Defects and Genetic Disorders

There are some tests that can be done before your baby is born. These are called prenatal tests and they check for certain birth defects or genetic disorders in your unborn baby.

What is a birth defect?

A birth defect is a health problem that a baby is born with. Birth defects can be mild or severe. Some only cause minor problems or can be treated. Others cause serious physical or mental handicaps.

What causes birth defects?

No one knows what causes most birth defects, but we do know that some can be caused by environmental factors, genetic factors, or both.

Environmental factors are things outside the baby, like drugs, alcohol, X-rays, and some infections in the mother. These are dangerous while you are pregnant because they can cause birth defects.

Genetic factors are inside the unborn baby, in its genes. Many things about us, like the color of our hair, eyes and skin, are controlled by our genes. Genes usually come in pairs, half from the mother and half from the father. If there are changes or "mistakes" in the genes, or if there are too many or too few genes, the baby can have a genetic disorder.

What is a genetic disorder?

A genetic disorder is a health problem caused by a mistake in a person's genes. There is no way to control which genes the unborn baby will get. Some genetic disorders are inherited, which means they run in the family. Others just happen and no one really knows why. All genetic disorders start before birth, but not all of them cause problems at birth. Some don't even begin to cause problems until the person is an adult. Like birth defects, some genetic disorders cause only mild problems while others cause severe mental or physical handicaps. ■

What Are Your Chances?

Everyone has a small chance of having a baby with a problem. Sometimes there are things in a woman's medical history or family history that increase the chances that her baby could have a birth defect or genetic disorder. If any of the sentences below fit you, your chances of having a baby with a problem are higher than normal.

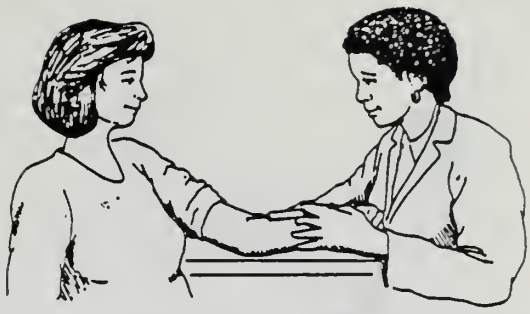
Medical History

- ☐ You have a medical problem (like diabetes, kidney disease, or a heart problem).
- ☐ You are 35 or older.
- ☐ You must take medicine for a health problem.
- ☐ You are on a special diet.
- ☐ You drink alcohol or use street drugs (like marijuana, crack or cocaine) while you are pregnant.
- ☐ You work with or have been exposed to dangerous chemicals or radiation (X-rays, for instance).
- ☐ You have been exposed to German measles (rubella), chicken pox, toxoplasmosis, or cytomegalovirus (CMV) while pregnant, or AIDS or genital herpes before you got pregnant.
- ☐ You have had two or more miscarriages.

Family History

- ☐ Someone in your family or in the baby's father's family was born with mental retardation or with a birth defect or genetic disorder.
- ☐ You have already had a baby with a birth defect, or a baby who was stillborn, or one who died soon after being born.
- ☐ You and the baby's father are blood relatives.
- ☐ You or the baby's father are from an ethnic group that is more likely to have a certain genetic disorder, such as:
  - *Ashkenazi Jews or French Canadians at risk for Tay-Sachs disease;*
  - *Blacks, Hispanics, or Cape Verdeans at risk for sickle cell conditions;*
  - *Caucasians at risk for cystic fibrosis;*
  - *Italians, Greeks or Asians at risk for thalassemia; or*
  - *Portuguese at risk for Joseph's disease.*

Remember: Even if more than one of these sentences fits you, it does not mean that your unborn baby will have a birth defect or genetic disorder. It just means your chances are higher than normal, and genetic counseling might be helpful to you. (If you want to learn more about genetic counseling, turn to page 12.) ■



MSAFP Screening Test

What is the MSAFP screening test?

MSAFP stands for Maternal Serum Alpha-FetoProtein (al-fuh-fee-toe-pro-teen). The baby growing in your uterus (womb) makes Alpha-FetoProtein, or AFP. Some of this AFP passes into your blood.

What is the MSAFP screening test for?

This test can tell you if you are more likely than normal to be carrying a baby with spina bifida (open spine), Down syndrome, or certain other birth defects. This is only a screening test, so it cannot tell you whether or not your unborn baby really has any of these problems. It can only tell you if the chances are higher than normal.

Who is the test for?

Doctors should offer this test to all pregnant women. It's up to you to decide whether or not you want it.

How is the test done?

A little blood is taken from your arm. The lab checks to see if the amount of AFP in it is high, low or normal. Most MSAFP results are normal.

When is the test done?

This test is done early in the 4th month of pregnancy (between 16 and 18 weeks). It can take one to two weeks for results to come back.

What if your MSAFP result is high?

If your result is high, it might mean that your pregnancy is further along than you thought, or that you're going to have twins. Sometimes it means that your unborn baby may have spina bifida or some other rare birth defect.

What if your MSAFP result is low?

If your result is low, it might mean that your pregnancy is not as far along as you thought. Sometimes it means that your unborn baby may have Down syndrome.

High or low results do not always mean that there is a problem with your unborn baby. Normal results do not always mean that your baby will be healthy. MSAFP is just a screening test, to help you and your doctor decide if you should think about having other tests done, like ultrasound or amniocentesis.

Is this test safe?

Yes. This test is safe for you and your unborn baby.

Does it hurt?

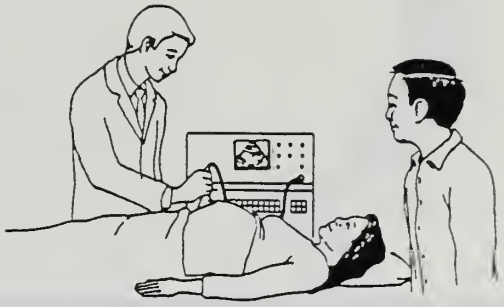
Most women say the needle feels like a pinch.

How much does the MSAFP screening test cost?

The test costs $35 to $50. Medicaid and most insurance companies will usually pay for this test. To be sure, check with the agency that pays for your health care.

What is the MSAFP Triple Test (also called AFP Profile or AFP Plus)?

This test is almost the same as the MSAFP screening test. The only important difference is that it is a better screening test for Down syndrome. ■



Ultrasound

What is ultrasound?

Ultrasound is a way to see your baby while it is still in your uterus (womb). It uses sound waves instead of X-rays to make pictures of the baby and the placenta (afterbirth). Sometimes ultrasound is called a "sonogram."

What is ultrasound for?

Ultrasound shows the size, shape and position of your baby inside you, so it can be used for many different reasons. Ultrasound pictures look fuzzy to most people, but doctors and specialists can tell a lot from them. They can see how your baby is growing. They can tell if you're going to have more than one baby. They can check the position of the baby and the placenta. If you have been bleeding, sometimes they can find out why. They can also check for some birth defects if your MSAFP result was high or low.

Ultrasound can show so much that doctors even use it to help with other tests, like amniocentesis. You should keep in mind, though, that some problems will not show up on ultrasound.

Who is it for?

Some doctors suggest ultrasound for all pregnant women to see how far along they are. Other doctors only suggest it if they think there might be a problem—for instance, if your MSAFP result was high or low.

How is it done?

Ultrasound can be done two ways. One way is to slide a small device, like a microphone, across your belly. The other way is to put another kind of microphone in the vagina. Both of these devices are attached to a machine that has a small black-and-white TV screen. Your unborn baby's picture shows up on this screen.

When is ultrasound done?

This test can be done any time during pregnancy. When it is used to check for certain birth defects, ultrasound is done early in the 4th month of pregnancy (between 16 and 18 weeks). You can usually get the results within a day.

Is ultrasound safe?

Most doctors consider it safe for you and your unborn baby.

Does it hurt?

Not at all. There are no needles. The microphone on your belly just slides across your skin. It can be a little uncomfortable, however, because you must hold your urine so you have a full bladder for this test. When ultrasound is done the other way, the device is gently guided inside your vagina. You do not have to have a full bladder for this test.

How much does ultrasound cost?

The test costs $150 to $500. Medicaid and most insurance companies will usually pay for this test. To be sure, check with the agency that pays for your health care. ■

Amniocentesis

What is amniocentesis?

Amniocentesis (am-nee-o-sen-tee-sis) is a test done on amniotic fluid, the water around the baby in the uterus (womb). Sometimes this test is called an "amnio" or a "tap."

What is this test for?

Amniocentesis can show if your unborn baby has Down syndrome or spina bifida. Sometimes it can show if your unborn baby has certain other birth defects or genetic disorders. However, some problems will not show up on amniocentesis.

Who is this test for?

Your doctor might suggest amniocentesis if:

- Your MSAFP test result was high or low.
- You have already had a child with a birth defect.
- A genetic problem runs in your family, or in the baby's father's family.
- You are 35 or older.

How is amniocentesis done?

A needle is carefully put through the skin on your belly and into your uterus to take out a little of the amniotic fluid. Doctors use ultrasound to see where to put the needle.

When is amniocentesis done?

This test is usually done early in the 4th month of pregnancy (between 16 and 18 weeks). Some doctors offer it earlier, at 12 weeks. It can take two to four weeks for results.

Is amniocentesis safe?

This test is almost always safe if it is done by an experienced doctor. Sometimes amniocentesis causes problems like cramping, bleeding, leaking amniotic fluid, or infection. There is also a small risk of poking the baby with the needle or causing a miscarriage, but these problems are very rare.

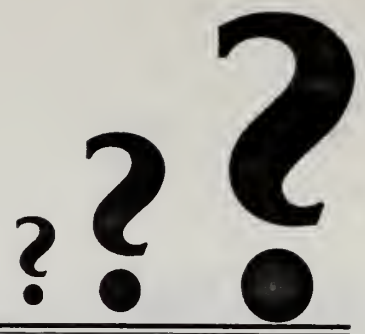
Does it hurt?

Most women say the needle feels like a pinch, and that it doesn't hurt as much as they thought it would.

How much does amniocentesis cost?

The test costs $500 to $1000. Medicaid and most insurance companies will usually pay for this test. To be sure, check with the agency that pays for your health care. ■

What If...?



Most prenatal test results are normal and most babies are born healthy. But what if ultrasound or amniocentesis shows that your unborn baby has a special problem? If that happens, there are some choices you can make.

For instance, you could:

Plan for special care

Your baby might need special care or treatment, like surgery, medicines, or a special diet. If you know what the problem is before your baby is born, you can plan ahead.

Plan for adoption or foster care

If you don't think you'll be able to take care of the baby yourself, you might want to consider adoption. There are parents who want to adopt children with special needs. If you're not sure adoption is right for you, foster care will give you more time to decide what to do.

End the pregnancy

If a test shows that the unborn baby has a problem, you might choose to have an abortion.

Which choice is best for you depends on your beliefs, your situation, what kind of problem the test turns up, and how serious the problem is.

If ultrasound or amniocentesis spots a problem, you should talk to doctors, specialists, and parents of children who have the same problem before you decide what to do. These are the people who can tell you the most about what to expect, so you can make the choice that's best for you.

You don't have to make a choice now. Just knowing what these choices are can help you decide whether or not you want to have prenatal tests. ■

Before You Decide

Any time a prenatal test is suggested, it's up to you to decide if you want it. Before you make up your mind, ask your doctor, nurse, midwife or genetic counselor these questions:

Why do you think I should have this test?

What can this test tell me?

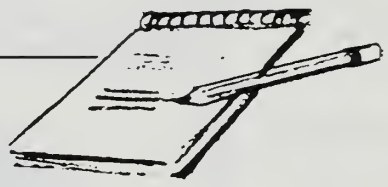
What are the risks of this test?

If the test finds a problem, how will it affect my baby?

If the test finds a problem, what can I do?

If you have any other questions about a test, ask them. The more you know, the easier it will be to make the decision that's best for you. ■

Notes



Genetic Counseling

Genetic counseling can help you if you are worried that your unborn baby has a special problem. Most women who get genetic counseling early in their pregnancies feel better after learning about all the choices they have.

When you go to a genetic counseling center, a trained genetic counselor will talk with you (and your partner, if he goes) about your health and any health problems that run in your families. The counselor will also discuss your chances of having a baby with a problem, and what prenatal tests can tell you. Genetic counseling can be done before or during pregnancy to help you decide whether or not to have any prenatal tests.

For some genetic disorders, you and your partner can have tests done even before you get pregnant. These tests can give you a better idea of your chances of passing the condition on to your children. A genetic counselor can explain both genetic and prenatal tests to you.

If you think genetic counseling will be helpful to you, ask your doctor, nurse or midwife to suggest a counselor, or call the center nearest you. If you want to know more, call:

Connecticut
State Genetics Services
Department of Pediatrics
University of Connecticut at Farmington
(203) 674-1465

Maine
Division of Maternal and Child Health
(207) 289-3311

Massachusetts
Massachusetts Department of Public Health
Genetics Program
(617) 727-5121 or 5125

New Hampshire
Division of Maternal and Child Health
(603) 271-4528

Vermont
Regional Genetics Center
(802) 658-4310

Rhode Island
Division of Family Health
(401) 277-2312

Other Resources

National Center for Education in Maternal and Child Health
38th and R Street NW
Washington DC 20057
(202) 625-8400

Spina Bifida Association of America
1700 Rockville Pike, Suite 250
Rockville, MD 20852
(301) 770-7222
1-800-621-3141

National Down Syndrome Congress
1800 Dempster Street
Park Ridge, IL 60088
(312) 823-7550
1-800-237-6372 (outside Illinois)

National Down Syndrome Society
666 Broadway
New York, NY 10012
1-800-221-4602

Remember, most babies are healthy!

This publication is available in Chinese, English, French, Khmer (Cambodian), Laotian, Portuguese, Spanish and Vietnamese.

For additional information about this booklet, contact:

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